

L Number	Hits	Search Text	DB	Time stamp
1	0	autosomal same hemochromatosis same mutation same ferroportin	USPAT; US-PGPUB; DERWENT	2003/07/31 13:18
2	2	"2002033119"	USPAT; US-PGPUB; DERWENT	2003/07/31 13:18
3	56375	mutations smae ferroportion same hemochromatosis	USPAT; US-PGPUB; DERWENT	2003/07/31 13:19
4	0	mutations same ferroportion same hemochromatosis	USPAT; US-PGPUB; DERWENT	2003/07/31 13:19
5	0	mutations same ferroportion	USPAT; US-PGPUB; DERWENT	2003/07/31 13:19
6	4	ferroportin same hemochromatosis	USPAT; US-PGPUB; DERWENT	2003/07/31 13:22

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TITLE: Autosomal-dominant **hemochromatosis** is associated with a missense **mutation** of the **ferroportin** 1 gene

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AB The present invention relates to **mutations** in the gene coding for **ferroportin** 1 assocd. with hereditary **hemochromatosis** and methods for the diagnosis of hereditary **hemochromatosis** based on the identification of such **mutations**. **Hemochromatosis** is a progressive iron overload disorder that is prevalent among individuals of European descent. It is usually inherited in an autosomal-recessive pattern and assocd. with missense **mutations** in HFE, an atypical major histocompatibility class I gene. Recently, the authors described a large family with autosomal-dominant **hemochromatosis** not linked to HFE and distinguished by early iron accumulation in reticuloendothelial cells. Through anal. of a large pedigree, the authors have detd. that this disease maps to 2q32. The gene encoding **ferroportin**, a transmembrane iron export protein, lies within a candidate interval defined by highly significant lod scores. The authors show that the iron-loading phenotype in autosomal-dominant **hemochromatosis** is assocd. with a nonconservative missense **mutation** in the **ferroportin** gene. This missense **mutation**, converting alanine to aspartic acid at residue 77 (A77D), was not seen in samples from 100 unaffected control individuals. The authors propose that partial loss of **ferroportin** function leads to an imbalance in iron distribution and a consequent increase in tissue iron accumulation.

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